MVK gene

mevalonate kinase

Normal Function

The *MVK* gene provides instructions for making the mevalonate kinase enzyme. This enzyme converts a substance called mevalonic acid into mevalonate-5-phosphate. This conversion is the second step in a pathway that produces cholesterol. The cholesterol is later converted into steroid hormones and bile acids. Steroid hormones are needed for normal development and reproduction, and bile acids are used to digest fats. Mevalonate kinase also helps to produce other substances that are necessary for certain cellular functions, such as cell growth, cell maturation (differentiation), formation of the cell's structural framework (the cytoskeleton), gene activity (expression), and protein production and modification.

Health Conditions Related to Genetic Changes

mevalonate kinase deficiency

At least 80 mutations in the *MVK* gene have been found to cause mevalonate kinase deficiency. There are two types of mevalonate kinase deficiency, which are differentiated by the severity of the signs and symptoms. The less severe of the two types is called hyperimmunoglobulinemia D syndrome (HIDS) and the more severe type is called mevalonic aciduria (MVA). Most of the *MVK* gene mutations that cause mevalonate kinase deficiency lead to changes in single protein building blocks (amino acids) in the mevalonate kinase enzyme. One mutation that replaces the amino acid valine with the amino acid isoleucine at position 337 in the enzyme (written as Val337Ile or V337I) is found in approximately 80 percent of people with HIDS. The V337I mutation has never been found in people with MVA.

Most *MVK* gene mutations lead to the production of a mevalonate kinase enzyme that is unstable and folded into an incorrect 3-dimensional shape, leading to a reduction of mevalonate kinase enzyme activity. The severity of the enzyme shortage (deficiency) determines the severity of the condition. People who have approximately 1 to 20 percent of normal mevalonate kinase activity typically develop HIDS. Individuals who have less than 1 percent of normal enzyme activity usually develop MVA. Despite this shortage of mevalonate kinase activity, people with mevalonate kinase deficiency typically have normal production of cholesterol, steroid hormones, and bile acids.

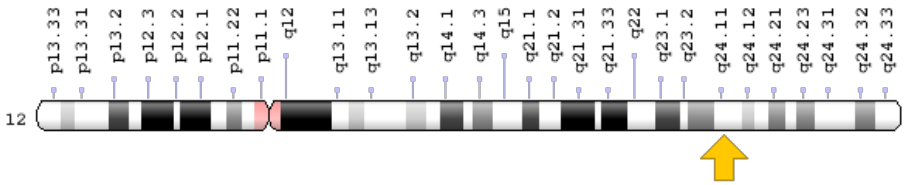
Some researchers believe the features may be due to a buildup of mevalonic acid, the substance that mevalonate kinase normally acts on. Other researchers think

that a shortage of the substances produced from mevalonic acid, such as those substances necessary for certain cellular functions, causes the fever episodes and other features of this condition. The exact mechanism that causes inflammatory reactions such as fevers, skin rashes, elevated immune system proteins, and many other features of mevalonate kinase deficiency is unclear.

Chromosomal Location

Cytogenetic Location: 12q24.11, which is the long (q) arm of chromosome 12 at position 24.11

Molecular Location: base pairs 109,573,285 to 109,597,270 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- KIME_HUMAN
- LH receptor mRNA-binding protein
- LRBP
- mevalonate kinase 1
- MK
- MVLK

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MVK%5BTIAB%5D%29+OR+%28mevalonate+kinase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- MEVALONATE KINASE
  http://omim.org/entry/251170

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MVK%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7530
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/4598
- UniProt
  http://www.uniprot.org/uniprot/Q03426

Sources for This Summary

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